A B S T R A C T

SEQUENCES

A method for detecting the presence or absence of twelve mutations in the cystic fibrosis transmembrane conductor regulator (CFTR) gene, which method comprises contacting sample genomic DNA from an individual in two separate reaction vessels with allele specific primer sets for (A) 1717-1 G>A, G542X, W1282X, N1303K, ΔF508(M), 3849+10kb C>T mutations and (B) the 621+1 G>T, R553X, G551D, R117H, R1162X and R334W mutations respectively, in the presence of appropriate nucleotide triphosphates and an agent for polymerisation, such that each diagnostic primer is extended only when the relevant mutation is present in the sample; and detecting the presence or absence of CFTR gene alleles by reference to the presence or absence of diagnostic primer extension product(s).